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(54) Title: DETECTION OF EPIGENETIC ABNORMALITIES AND DIAGNOSTIC METHOD BASED THEREON

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Sch74-E52m/Sch74-E51m  CAGCTCACTGCAACCTCCGCC-TCCTGGATTCAAGC-GATTTTCCCGCCTTAG-CCTCCTGAGTAACTGGGACTAGAGGCAGGTACCACCACGCCCAGCTAATTTTT-GTATT
BD43-E78m/BD43-E83m    CGGCTCAATGCAACCTCAGCC-TCCTGGGTTCAAGC-AATTCTCCTGTCTCAG-CCTCCCGAGTAGCTGGGATTACAGGCACATGCCACCATGCCCAACTAATTTTT-GTATT
BD43-E78m              CGGCTCAATGCAACCTCAGCC-TCCTGGGTTCAAGC-AATTCTCCTGTCTCAG-CCTCCCGAGTAGCTGGGATTACAGGCACATGCCACCATGCCCAACTAATTTTT-GTATT
BD34-D19M              TGGCTCACTGTAACCTCTGCC-TCCTGGGTTCAAGT-AATTCTCCTGTCTCAG-CCTCCTGAGTAGCTAGGATTACTGGTGCCCGCCACCATGCCCGGCAAATTTTT-GTATT
BD34-E62m              TGGCTCACTGTAACCTCTGCC-TCCTGGGTTCAAGT-AATTCTCCTGTCTCAG-CCTCCTGAGTAGCTAGGATTACTGGTGCCCGCCACCATGCCCGGCAAATTTTT-GTATT
BD43-15m               TGGCTCACTGCAACCTCTACC-TCCTGAGTTCAAGC-TCTTCTCCTGCCTCAA-CCTCCAGAGTAATTGTGATTACAGGTGCCTCCCACCACACCAGGCTAATTTTT-GTATT
Ctr157-E5m             CAGCTCACTGCAACCTCCATT-TCCTGGGTTCAAGC-GATTCTCCTGCCTCAG-CCTCCGGAGTAGCTGGGACCACAGACGTGTGCCACCATGCCTGGGTAATTTTC-ATATT
BD43-RevE7m            CAGCTCACTGCAACCACCACC-TCCCAGGTTCAAGT-GATTATCCTGCCTCAG-CCTCCCGAGTAGCTGGGATTACAGATGCCCACCAACACACCAGGCTAATTTTTTGTATT
BD43-RevE77m           CAGCTCACTGCAACCACCACC-TCCCAGGTTCAAGT-GATTATCCTGCCTCAG-CCTCCCGAGTAGCTGGGATTACAGATGCCCACCAACACACCAGGCTAATTTTTTGTATT
BD34-A14M              CAGCCCAGTGCAAGCTCCGCC-TCCCAGGTTCACGT-CATTCTCCTGCCTCAG-CCTCCCGAGTAGCTGGGACTACAGGCGCCCGCCACCACGCCCAGCTAATTTTTTGTATT
Ctr157-E3m             CGGCTCACTGCAAGCTCCGCC-TCCCGGGTGCACGC-CATTCTCCTGCCTCAG-CCTCCCGAGTAGCTGGGACTACAGGCGCCCGCCACCACGCCCGGCTAATTTTTTGTATT
Sch74-E318m            TGGCTCACTGCAACCTCCGCC-TCCCAGGTTCAAGC-AATTCTCCTGCCTCAG-TCTCCCGAGTAGCTGGGACTACCGGCGAGTGCTACCATGCCTGCGTAATTTTTTGTACT
Sch74-E318_m           TGGCTCACTGCAACCTCCGCC-TCCCAGGTTCAAGC-AATTCTCCTGCCTCAG-TCTCCCGAGTAGCTGGGACTACCGGCGAGTGCTACCATGCCTGCGTAATTTTTTGTACT
Ctr157-E4m             TGGCTCACTGCAACCTCCGCC-TCCCAGGTTCAAGC-AATTCTCCTGCCTCAG-TCTCCCGAGTAGCTGGGACTACCGGCGAGTGCTACCATGCCTGCGTAATTTTTTGTACT
BD43-E79m              CAGCTCACTGCAACCTCCGTT-TCCCAGGTGCAACC-GATTCTCCTGCCTCAGACCTCTGAAGCGGCTGGGACTACAGGTGCCTGCCACCTCACCCGGCTAATTTTT-GTATT
Ctr157-E6m             CAGCTCACCACAACCTCCGCC-TCCTGGGTTCCAGC-GATTCTCCTGCCTCGG-CCTCCCAAGTAGCTGGGATTACAGGCACGCACCAATACACCTGGCTAATTTTG--TATT
Ctr157-E6m             CAGCTCACCACAACCTCCGCC-TCCTGGGTTCCAGC-GATTCTCCTGCCTCGG-CCTCCCAAGTAGCTGGGATTACAGGCACGCACCAATACACCTGGCTAATTTTG--TATT
Ctr150-RevE169m        CAGCTCTCCACAACCTCCGCCATCGTGGGTTCCAGCAGATTCTCCTGCCTCGG-CCTCCCAAGTAGCTGGGAATACAGGCACGCTCCAATACACCTGGCTAATTATG--TATT
Sch56-E283m            CAGCTCACCGCAACCTTTGCC-TCACGGGCTCAAGT-GATTCTCATGCTTGAT-CCTACCAAGTAGCTGGGATTACAGGCACATGCCATCATGCTGAGCTAACTTTG-GTATT
Sch56-r-37m            CAGCTCACCGCAACCTTTGCC-TCACGGGCTCAAGT-GATTCTCATGCTTGAT-CCTACCAAGTAGCTGGGATTACAGGCACATGCCATCATGCTGAGCTAACTTTG-GTATT
Sch56-E32m             CACGTCACTGTAATGTCCATC-TCCCGGGTTCAGGT-GATTCTCCTGCCCCAG-CCTCCTGAGTAGCTG----TACAGGCGTGCACCACCATGCCCGACTAATTTTT-GTACT
Ctr150-E166m           CGGCCCACTGCAACCTCCGCC-TCCCGGGTGCAAGC-AGTTCTCCTACCTCAG-CCTCCTGAGTAGCTAGGATTACAGG----------CACACCTGGCTAATTTTG--TGGT
Ctr150-E49m            CGACTCATTGCAACCTCTGCC-TCCTGGGTTTAAGC-CGTTCTCATGCCTCAGCCTCCCGACGTAGCTGGGATTATAGGCATGCGCCACCACCCCCAGCTAATTTTT-GTATT
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(57) Abstract: The present invention provides a method of detecting an epigenetic abnormality associated with a disease. The method comprises identifying, within a eukaryotic genome, a locus having a hypomethylated sequence specific for the disease and an endogenous multi-copy DNA element. The method can also comprise separate steps of identifying a disease-specific hypomethylated sequence and identifying an endogenous multi-copy DNA element, where the steps may be performed in any order, so long as a locus is identified that has both a disease-specific hypomethylated sequence and an endogenous multi-copy DNA element. The disease-specific hypomethylated sequences detected in accordance with the present invention indicate putative regions of epigenetic dys-regulation and indicate aberrantly regulated nucleic acid sequences that may cause or predispose a patient to disease, such as, but not limited to, Huntingdon s disease, cancers, diabetes, schizophrenia, or bipolar disorder.

WO 03/104487 A2